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Media Statement

WA RESEARCHERS AIM TO FURTHER BOOST BONE FINDINGS

WA scientists are looking for volunteers for further research that has already delivered an important discovery which could uncover new treatments for chronic bone diseases.

The study investigating the genetic causes of Paget's disease of the bone (PDB) – a painful condition that causes bone inflammation and deformity – has revealed a new mutation in a gene linked to the disease. The mutation was detected in a WA patient.

The research was conducted by Western Australian Institute for Medical Research (WAIMR) PhD student Sarah Rea and supervised by the head of Sir Charles Gairdner Hospital's Department of Endocrinology & Diabetes, Clinical Associate Professor John Walsh, WAIMR Associate Professor Tom Ratajczak and University of WA Associate Professor of Orthopaedic Surgery Jiake Xu.

"The new genetic mutation we've found essentially 'chops off' the back end of a particular protein important for controlling bone loss – causing the protein function to be switched on continually, sparking excessive bone loss and very severe Paget's," said A/Prof Ratajczak.

"This discovery is providing many new insights and the ultimate aim would be to look at ways of turning off this protein function, which could allow for the creation of fresh treatments for this terrible condition."

A/Prof Walsh said the finding was also generating hope of treating other conditions.

"We believe this genetic mutation could also allow us to learn more about other bone diseases, including cancers of the bone, and may even prove useful in developing new therapies for those conditions," he said.

"We are also excited about the discovery of another new mutation we have detected in the same gene, but in a different area of it, which appears to cause milder cases of Paget's."

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A/Prof Ratajczak said the findings were only possible through collaboration between clinical and scientific teams – and with the help of almost 300 WA patients to date.

“In order to continue to investigate our discoveries and look for more clues for treating these devastating bone diseases, we’re hopeful more West Australians affected by Paget’s will put up their hands to help us put together the pieces of the puzzle,” he said.

Potential volunteers should call Lynley Ward on (08) 9346 2370.

The research was assisted by a \$15,000 grant from Arthritis Australia and has been published in the Journal of Bone & Mineral Research.

PAGET’S BACKGROUND

In PDB, the bones become weakened, soft and abnormally enlarged. Any part of the skeleton can be affected, but the most common sites include the skull, spine, pelvis, thigh-bone, shin and the bone of the upper arm.

The chronic disease affects up to 4% of individuals over the age of 55. It is more common in men than women.

As well as a genetic component, there is an environmental component to PDB, but what that is remains unknown.

In 95 per cent of cases, a person with Paget’s disease presents no symptoms. In other cases, the symptoms may develop gradually over a long period of time, or can quickly become severe and debilitating. Pain is often caused by the enlarged bone putting pressure on surrounding nerves.

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